

Claims

1. A genetic reference standard comprising at least one human genetic reference sequence cloned into a non-mammalian animal cell line.

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2. The genetic reference standard of claim 1 wherein the animal cell line is an avian cell line.

3. The genetic reference standard of claim 2 wherein the cell line is a chicken (*Gallus* spp.) cell line.

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4. The genetic reference standard of any preceding claim wherein the cell line is a B-cell line.

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5. The genetic reference material of claim 3 wherein the chicken cell line is the chicken DT40 cell line.

6. A genetic reference standard according to any preceding claim wherein the at least one human genetic reference sequence is cloned into a dispensable region of the cell's genome.

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7. A genetic reference standard according to any preceding claim wherein the at least one human genetic reference sequence is cloned into a non-expressed region of the cell's genome.

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8. A genetic reference standard according to any preceding claim wherein the cloned cell line is diploid with respect to the human genetic reference sequence.

9. A genetic reference standard according to any preceding claim wherein the at least one human genetic reference sequence is a plurality of human genetic reference sequences.

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10. A genetic reference standard according to any preceding claim wherein the or each human genetic reference sequence is not a functional chromosome.

11. A method of detecting a genetic variant in a sample containing human DNA comprising:

5 performing a test, responsive to DNA sequence, on said sample;

10 performing the same test on a reference sample embodying the genetic variant to be detected;

comparing the test results obtained from said sample and said reference sample to determine the presence or absence of said genetic variant;

characterised in that said reference sample is a genetic reference standard according to
10 any preceding claim.